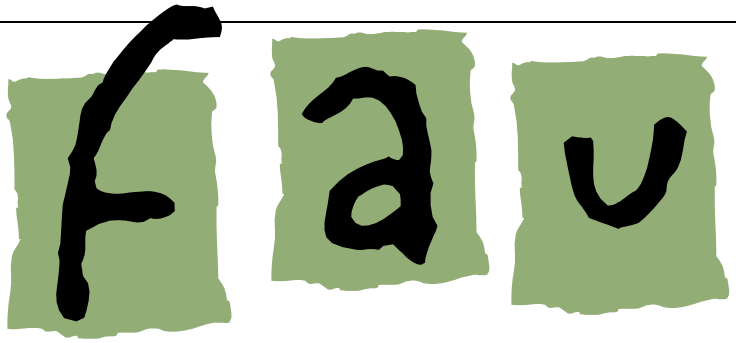


Unidentified Hemoglobin Traits

Each year thousand of infants born in the United States are detected with unidentified hemoglobin variants through newborn screening. Most of these infants are heterozygotes, with the quantity of adult hemoglobin (Hb A) being equal to or greater than the quantity of unidentified hemoglobin variant (Hb U). Mutations in the α-, β-, or γ-globin genes may cause these variants.

Most unidentified hemoglobin variants have no clinical or hematological consequence, but a few may show altered oxygen affinity or be chemically unstable. Some unidentified hemoglobin variants can cause sickle cell disease when co-inherited with hemoglobin S, but most unidentified hemoglobin variants have no significant genetic implications.

With over 600 structural hemoglobin variants, and limited reference laboratory capacity in the United States, the majority of unidentified hemoglobin variants identified by screening cannot be definitively identified. While the overwhelming number of unidentified hemoglobin variants has no clinical or genetic significance, uncertainty about the identity of variants may lead to frustration and anxiety for families and health care providers.

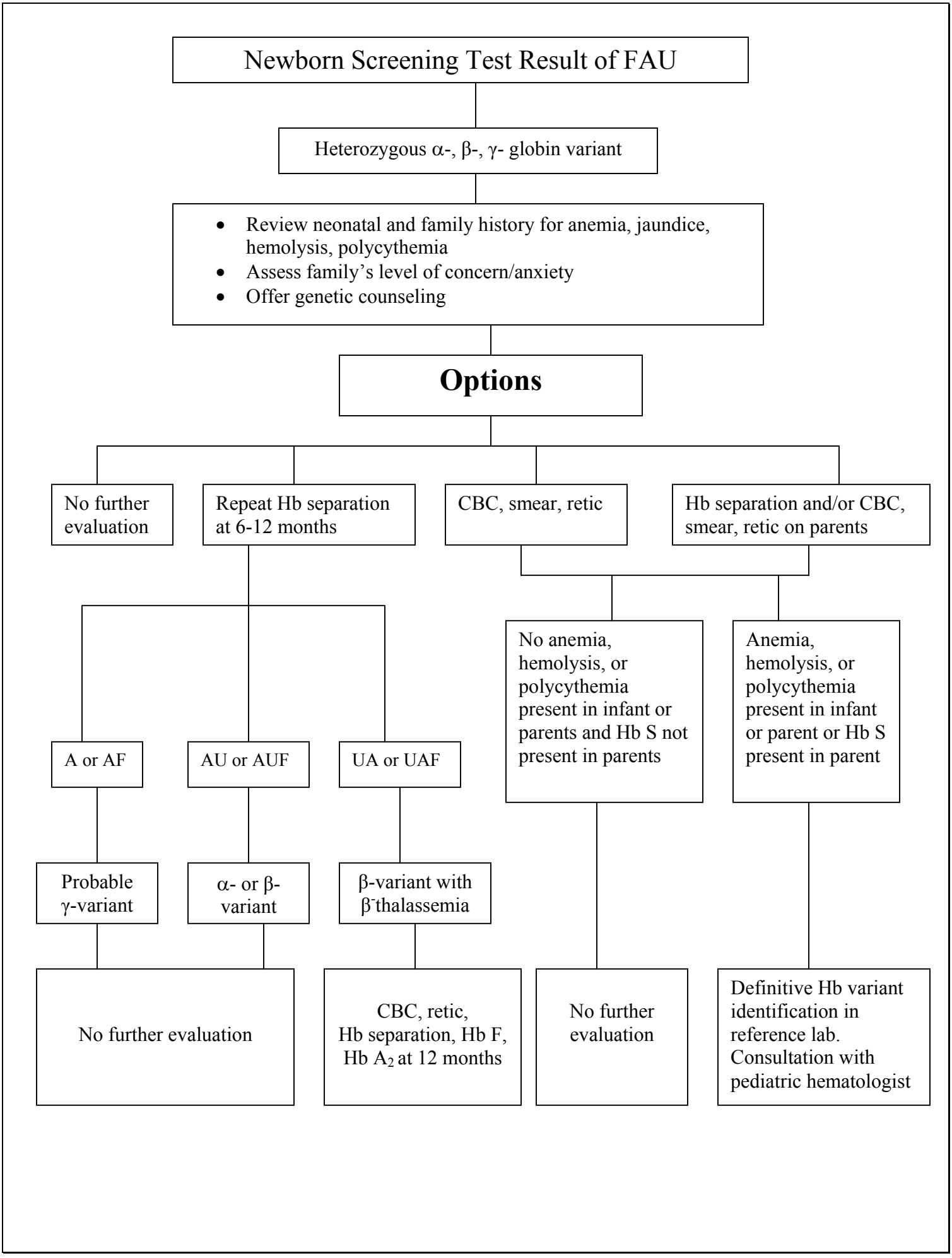
The algorithm, found on the back of this handout, is intended to provide a common-sense approach that should limit laboratory expense and reserve definitive hemoglobin variant identification in reference labs for situations where definitive identification is needed for specific clinical or genetic concerns in a given family.

Adapted from Sickle cell disease in children and adolescents: Diagnosis, guidelines for comprehensive care, and care paths and protocols for management of acute and chronic complications; Lane, P.A., Buchanan, G.R., Hutter, J.J., Austin, R.F., Brittion, H.A., Rogers, Z.R., et al.

Newborn Screening Program

Utah Department of Health
44 N Medical Dr
Salt Lake City, Utah 84114
Phone (801) 584-8256
http://health.utah.gov/newbornscreening

Promote Prevent Protect
Utah Department of Health


Newborn Screening Test Result of FAU
Heterozygous α-, β-, γ- globin variant
Review neonatal and family history for anemia, jaundice, hemolysis, polycythemia
Assess family's level of concern/anxiety
Offer genetic counseling
Options
No further evaluation
Repeat Hb separation at 6-12 months
CBC, smear, retic
Hb separation and/or CBC, smear, retic on parents
A or AF
AU or AUF
UA or UAF
No anemia, hemolysis, or polycythemia present in infant or parents and Hb S not present in parents
Anemia, hemolysis, or polycythemia present in infant or parent or Hb S present in parent
Probable γ-variant
α- or β-variant
β-variant with β-thalassemia
No further evaluation
CBC, retic, Hb separation, Hb F, Hb A2 at 12 months
No further evaluation
Definitive Hb variant identification in reference lab. Consultation with pediatric hematologist